Sue is a 31-year-old woman whose father died at the age of 53 of hemophilia, an X-linked recessive condition. Sue's 40-year-old sister, Ann, has a 15-year-old son named Tom who has hemophilia. Sue's father was of Scottish descent and her mother is Irish. Sue is now pregnant. When Sue and her husband Jon (34 years old) were seen in the genetics clinic, their genetic counselor told them that Sue is an obligate carrier of hemophilia, given her family history. Each son she has faces a 50% chance of being affected, and each daughter has a 50% chance of being a carrier. Sue and Jon decide to do prenatal diagnostic testing for hemophilia if ultrasound shows that Sue is carrying a boy.

Prenatal diagnostic genetic testing is most accurate when the fetus is tested for a known familial mutation. Sue finds out from Ann that a mutation was identified when DNA testing was done on her nephew, Tom. Sue and Jon's genetic counselor gets a signed release of records from Ann and is then able to get a copy of Tom's DNA test results.

By ultrasound examination, Sue's baby appears to be male, so amniocentesis is performed, and the amniotic fluid is sent to the same laboratory that tested Tom. The laboratory needs to know the relationships between the members of the family in order to interpret the test results, so the doctor includes the pedigree shown below with the samples.

COMMON STANDARD PEDIGREE SYMBOLS AND ABBREVIATIONS*

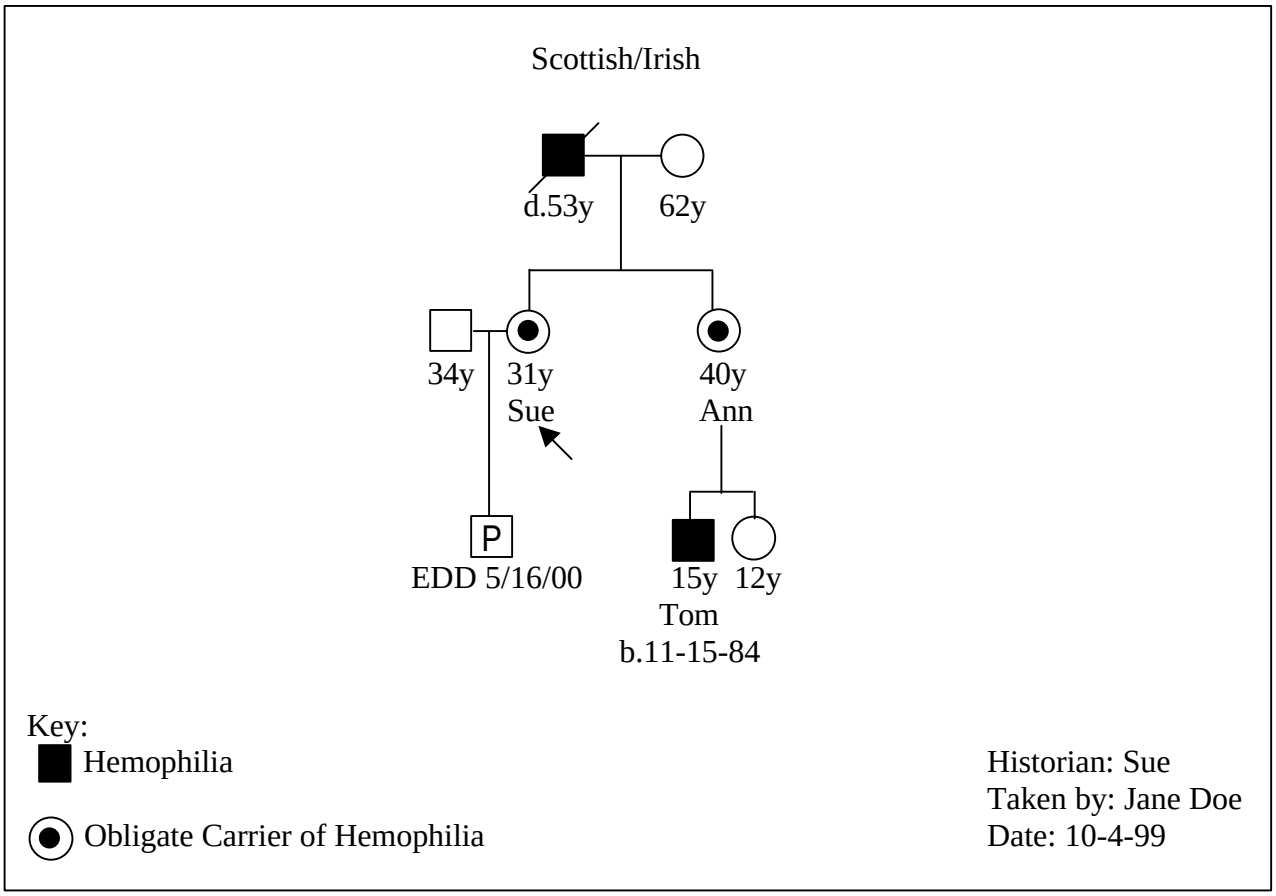
	Male	P	Pregnancy
	Female	d.	died
	Gender Unknown	b.	born
	Affected Individual	y	years
	Deceased Individual	EDD	Estimated Date of Delivery

 Consultand (the person who presents for services)

***** Note: More symbols and abbreviations are shown under "What is a Genetic Consultation"

(Scroll down for pedigree)

Pedigree used to provide clinical information to the genetic testing laboratory*



*****Note: A more detailed family history is shown under "What is a Genetic Consultation"